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Current Estimate of Down Syndrome Population Prevalence in the United States

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Abstract

Objective—To calculate a reliable estimate of the population prevalence of Down syndrome in the US.

Study design—The annual number of births of infants with Down syndrome were estimated by applying published birth prevalence rates of Down syndrome by maternal age to US data from the Centers for Disease Control and Prevention for the years for which births by maternal age were available (1940–2008). Death certificate data for persons with Down syndrome were available for the years 1968–2007. We estimated the number of people with Down syndrome on January 1, 2008, using a life table approach based on proportions of deaths by age. Monte Carlo sampling was used to create 90% uncertainty intervals (UIs) for our estimates.

Results—We estimated the January 1, 2008, population prevalence of Down syndrome as approximately 250 700 (90% UI, 185 900–321 700) based on proportions of deaths by age from the most recent 2 years (2006–2007) of death certificate data. This estimate corresponds to a prevalence of 8.27 people with Down syndrome per 10 000 population (90% UI, 6.14–10.62).

Conclusion—Our estimate of Down syndrome prevalence is roughly 25%–40% lower than estimates based solely on current birth prevalence. The results presented here can be considered a starting point for facilitating policy and services planning for persons with Down syndrome.

Down syndrome is one of the most common birth defects in the US with approximately 6000 births annually, resulting in an estimated birth prevalence of 14 per 10 000 live births.[1–3] Persons with Down syndrome have experienced significant increases in life expectancy over the past few decades, owing to reduced institutionalization and improved access to medical care, such as surgical intervention for congenital heart defects.[4–8]

The authors declare no conflicts of interest.

Although this increase in life span points to an increasing number of people living with Down syndrome over the past few decades, very little is known about the actual prevalence of Down syndrome. National population-based prevalence estimates of birth defects based on data from birth defects surveillance programs in the US were first published in 2006 (using data for birth years since 1999) and currently are based on only approximately one-third of US births.[1,2] Furthermore, registry systems do not routinely provide follow-up after birth, so that deaths of persons with Down syndrome must be determined from separate sources.[3,9] As a result, there is currently no reliable estimate of the number of people living with Down syndrome in the US. Such an estimate is essential for prioritizing Down syndrome research and planning services for persons with Down syndrome.

The National Down Syndrome Society[10] and rightdiagnosis.com[11] report US population estimates of persons with Down syndrome as 400 000 and 340 000, respectively, based on estimated birth prevalence. To our knowledge, these estimates rely on assumptions of a fixed US population size, similar frequency of death in persons with Down syndrome as in the general population, and a consistent prevalence of Down syndrome over time.

US studies that have attempted to estimate the number of persons with Down syndrome have done so for infants or children and adolescents, focusing on data from birth defects surveillance systems. In a study of data from birth defects surveillance systems from 10 US regions for 1979–2003, Shin et al[9] estimated a prevalence of Down syndrome of 10.3 per 10 000 persons aged 0–19 years in 2002. A separate study using data from Atlanta's birth defects surveillance system estimated a prevalence of 8.3 per 10 000 persons aged 0–19 years in 2003.[3] In both cases, the authors focused on children and adolescents, because the majority of birth defects surveillance systems are less than 20 years old, which limited the ability to estimate the numbers of older individuals with Down syndrome.[12]

The majority of US demographic studies of Down syndrome have focused either on birth prevalence using birth defects surveillance systems or on mortality data derived from death certificates. Studies have identified substantial differences in birth prevalence across states, birth defects surveillance systems, and time. Even though a lower average estimate was reported for the 1980s–1990s than for 1999–2006 (10.61 [range, 8.67–14] per 10 000 births[3,9,13–16] vs 12.45 [range, 9.35–14.47] per 10 000 births[1–3,9,13]), the ranges overlap. Most recently, Parker et al[1] studied the prevalence of Down syndrome births in data from 24 birth defects programs accounting for 32.3% of total US births, and estimated it as 14.47 per 10 000 live births. Previous mortality studies have been based on death certificate data from the Centers for Disease Control and Prevention (CDC), in which Down syndrome was listed on death certificates as either a cause or comorbidity.[5,17] These studies have shown a dramatic increase in median age at death over the past few decades, likely related to advances in medical care and reduced institutionalization.[4,5,17]

A recent study by de Graaf et al[18] estimated the prevalence of Down syndrome as 13.1 per 10 000 in Ireland, 7.7 per 10 000 in The Netherlands, and 6.1 per 10 000 in England/Wales. In their study, prevalence was estimated using maternal age and Down syndrome risk along with Down syndrome mortality rates derived from multiple studies, owing to a lack of

comprehensive registry data.[18] In the present study, we adopted a similar strategy to estimate the current number of persons with Down syndrome in the US.

The purpose of this study was to estimate the prevalence of Down syndrome in the US on January 1, 2008, using estimates of annual births of infants with Down syndrome over the past century. A Monte Carlo sampling approach was used to estimate the uncertainty in our population size estimates.

Methods

We estimated the number of persons with Down syndrome as of January 1, 2008, using data on live births of infants with Down syndrome each year for 1909–2007, by applying published birth prevalence estimates of Down syndrome by maternal age[19] to counts of births by maternal age from US national birth records compiled by the CDC[20,21] and age-specific mortality proportions from US national death certificate data for 1968–2007.[22,23] US birth data by maternal age group for 1940–2008 and total US births for 1909–1939 were available from the CDC and used to estimate births by maternal age. We used data on total US births by maternal age group for 1909–2008 to estimate the number of births of infants with Down syndrome using estimates of total birth prevalence of Down syndrome by maternal age. These data were from publicly available datasets and do not require institutional review board review. These prevalence estimates by maternal age were based on data derived from many countries, which have been shown to be relatively consistent across countries.[19,24] Although the estimates of total birth prevalence of Down syndrome by maternal age are internationally consistent, the live birth prevalence can differ significantly among countries owing to differences in maternal age composition and in pregnancy termination rates. Thus, the estimates of total births of infants with Down syndrome by year for 1909–2007 were further adjusted for pregnancy terminations. The adjustment for pregnancy terminations was estimated from the prevalence difference between live births plus pregnancy terminations versus live births alone reported by Parker et al (658 live births plus pregnancy terminations vs 575 live births).[1] It was applied uniformly across 1980–2007, because termination rates were similar in the 1980s and 1990s.[25] Data on number of deaths of persons with Down syndrome by year and age were available from the CDC for 1968–2007, using death certificate data in which Down syndrome was listed as either a direct cause or a comorbid condition. These data, along with age-specific US deaths for 1968–2007, total US deaths by age group for 1900–1998, and age-specific death rates of persons with Down syndrome were used to estimate age-specific mortality of persons with Down syndrome for 1900–1967. Age-specific deaths of persons with Down syndrome for 1968–2007 and the total live births of infants with Down syndrome per year for 1909–2007 were used to estimate the January 1, 2008, population of persons with Down syndrome.

All data formatting and analysis was conducted in R.[26] Because the population size estimates were derived and adjusted by several different sources and measures, we estimated uncertainty in our birth, death, and final population size estimates using Monte Carlo sampling.[27] We report the median and 90% uncertainty interval (UI; 5th and 95th percentiles) from 10 000 Monte Carlo iterations.

Results

The results presented here are intended to provide a general overview of the estimated number of persons with Down syndrome in the US as of January 1, 2008.

Estimation of Annual Live Births of Infants with Down Syndrome for 1909–2008

Because birth defects surveillance systems are fairly new and limited to specific US regions, we adopted the approach of de Graaf et al[18] for estimating annual births of infants with Down syndrome by applying maternal age-specific birth prevalence of Down syndrome to annual numbers of live US births.[19,24] Data on live births by maternal age group were available for 1940–2008, and data on total US live births were available for 1909–1994, from which we estimated the number of live US births per maternal age group for 1909–1939. We estimated the total number of births of infants with Down syndrome using birth rates per 1000 live births by 5-year maternal age groups reported by Carothers et al.[19] Figure 1, A shows that the number of live births with Down syndrome, which parallels the total number of US births, increased in the 1950s–1960s, followed by a decline in the 1970s, and then a steady increase since the 1980s.

Estimation of Age-Specific Annual Deaths of Persons with Down Syndrome for 1900–2007

Data on age-specific US deaths overall and of persons with Down syndrome were available for 1968–2007, and data on US deaths by 10-year age group were available for 1900–1968. Annual deaths in persons with Down syndrome before 1968 were estimated using the fraction of deaths associated with Down syndrome in the first 2 years of available data (1968–1969) along with total US mortality data for 1909–1967. Mortality for 1999–2007 was adjusted from the *International Statistical Classification of Diseases and Related Health Problems, 10th Revision* to the *9th Revision* using a comparability ratio[28] to produce estimated age-specific deaths of persons with Down syndrome by year for 1909–2007.

Even though the number of US deaths has been increasing since 1909, the deaths of persons with Down syndrome peaked in 1918, followed by a decline through 1980 (Figure 1, B). The difference in mortality curves between the US population and persons with Down syndrome can be attributed mostly to the increasing number of persons with Down syndrome surviving beyond age 45 years. Figure 1, B shows that the number of deaths per year has been increasing in the US population, and the number of deaths of persons with Down syndrome has been decreasing for most of the past century owing to a rapid increase in life span that started in the early 1970s (Figure 2). Figure 1, B also shows that the number of deaths of persons with Down syndrome began to increase in the 1990s, likely related to stabilization of the life span increase.

The life expectancy of persons with Down syndrome increased relatively slowly from 1900 to 1960 (by 89%) but rapidly from 1960 to 2007 (by 456%). The median overtook the mean, indicating a shift in distribution skew, in 1985 (Figure 2).

Estimation of the January 1, 2008, Population of Persons with Down Syndrome Based on Proportion of Deaths

Because death certificate data are known to reflect underreporting of chronic conditions, especially in persons with Down syndrome,[5,17] we used a life table approach to estimate the January 1, 2008, population size. This approach relied on annual proportions of deaths by age at a specific time point rather than a cohort life table approach, where proportions of deaths are specific to each birth cohort.[29] We estimated the January 1, 2008, population size using proportions of deaths by age obtained from the 2 most recent years of death certificate data (2006–2007) as 250 700 (90% UI, 185 900–321 700) (Figure 3). As a validation exercise, we applied the same method to estimate the total US population for January 1, 2008, and found it to be underestimated by approximately 14.6% (estimated size, 258 832 900 [90% UI, 258 750 300–258 915 700], compared with the US Census Bureau figure of 302 977 400). A 14.6% underestimate of the prevalence of Down syndrome suggests an actual population of approximately 293 600.

Discussion

Given the increased life expectancy of persons with Down syndrome and their associated health risks, a reliable estimate of current population size is needed to inform decisions on providing services to this group. We present such prevalence estimates based on birth and mortality data from the US along with estimated Down syndrome birth prevalence rates by maternal age. We estimate that on January 1, 2008, approximately 250 700 persons with Down syndrome (90% UI, 185 900–321 700) were living in the US. Validating this approach by applying the same methods to the total US population resulted in a January 1, 2008, population estimate approximately 14.6% lower than the actual size; however, a discrepancy of 14.6%in our estimated number of individuals with Down syndrome on January 1, 2008, is well within our UI.

Previous US studies have used birth certificate data[13,30,31] or data from birth defects surveillance systems[1–3,13] to estimate the prevalence of Down syndrome among births in the US population.[1–3,9] Our 1979–2006 estimates of the live birth prevalence of Down syndrome were approximately 16% larger than those presented elsewhere.[1,19–24,28] The mortality data analyzed here come from death certificate data derived from the same national registry used by other US studies on mortality in Down syndrome.[5,17] As a result, our estimates of mortality by age are nearly identical.[5,17]

Comparing our modern survival estimates with results from European studies yields some interesting findings. de Graaf et al[18] performed a comprehensive literature analysis of survival of persons with Down syndrome and developed a life expectancy model.[18] Although our median life expectancy results agree well for data from 1983–2000 (during which median life expectancy increased from 25 to 50 years in the US population and from24 to 52 years in the European studies), the earlier average life expectancy estimates from 1930–1980 reported by de Graaf et al were notably higher.[18] For example, de Graaf et al modeled a life expectancy of 16 years and reported literature estimates for 1963 life expectancy of 28 years, compared with 9.2 years in the present study. Their estimates for 1970 were 30 years (modeled estimate) and 35 years (reported estimate), compared with our

estimate of 12.6 years. Interestingly, it appears that the average life expectancy of persons with Down syndrome was approximately 50% lower in the US versus Europe from about 1960 to 1980.

Because there are no population prevalence estimates for Down syndrome in the US, we again compared our results with those of European studies. Our estimated prevalence of Down syndrome for 2008 was 8.27 (90% UI, 6.14–10.62) per 10 000 population, close to the estimated prevalences in The Netherlands (7.7 per 10 000) and England/Wales (6.1 per 10 000).[18]

A major limitation of our analysis is the underreporting of the diagnosis of Down syndrome in death certificate data, which caused us to use only proportions of age-specific deaths rather than actual numbers to estimate the population of persons with Down syndrome.[5,17]

At 250 700, our estimate for the prevalence of Down syndrome in the US is roughly 25%-40% below online estimates typically determined solely from birth rates (340 000[10] and 400 000[11]). A careful projection of Down syndrome prevalence through the year 2100 requires expert elicitation of determinative factors, such as future survival rates of persons with Down syndrome, pregnancy terminations, and policy or societal changes that could affect birth rates in older women. Nonetheless, a rough estimate of Down syndrome prevalence in 2100, assuming that prevalence increases uniformly with the increasing US population, would be 486 400–975 200 based on the middle and high projections of the US population size from the US Census Bureau. Given that the average age at death of persons with Down syndrome has increased dramatically since 1970, these projections could underestimate the prevalence for 2100.

This analysis estimated the annual US live births of persons with Down syndrome for 1909–2007, estimated US age-specific annual deaths of persons with Down syndrome for 1909–2007, and used this information to estimate the size of the US population of persons with Down syndrome using proportion of deaths. Existing estimates do not consider the shorter life expectancy for individuals with Down syndrome or the varying prevalence of Down syndrome over time. Our results can be used to facilitate planning for medical care and other services for persons with Down syndrome. We propose evaluating expert opinions to project the future prevalence of Down syndrome in the US.

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The findings and conclusions in this report are those of the authors and do not necessarily represent the official position of the Centers for Disease Control and Prevention. This work was performed when E.M. was a faculty member of the University of Colorado and does not represent a work product, conclusions, or policies of the March of Dimes.

Glossary

CDC	Centers for Disease Control and Prevention
UI	Uncertainty interval

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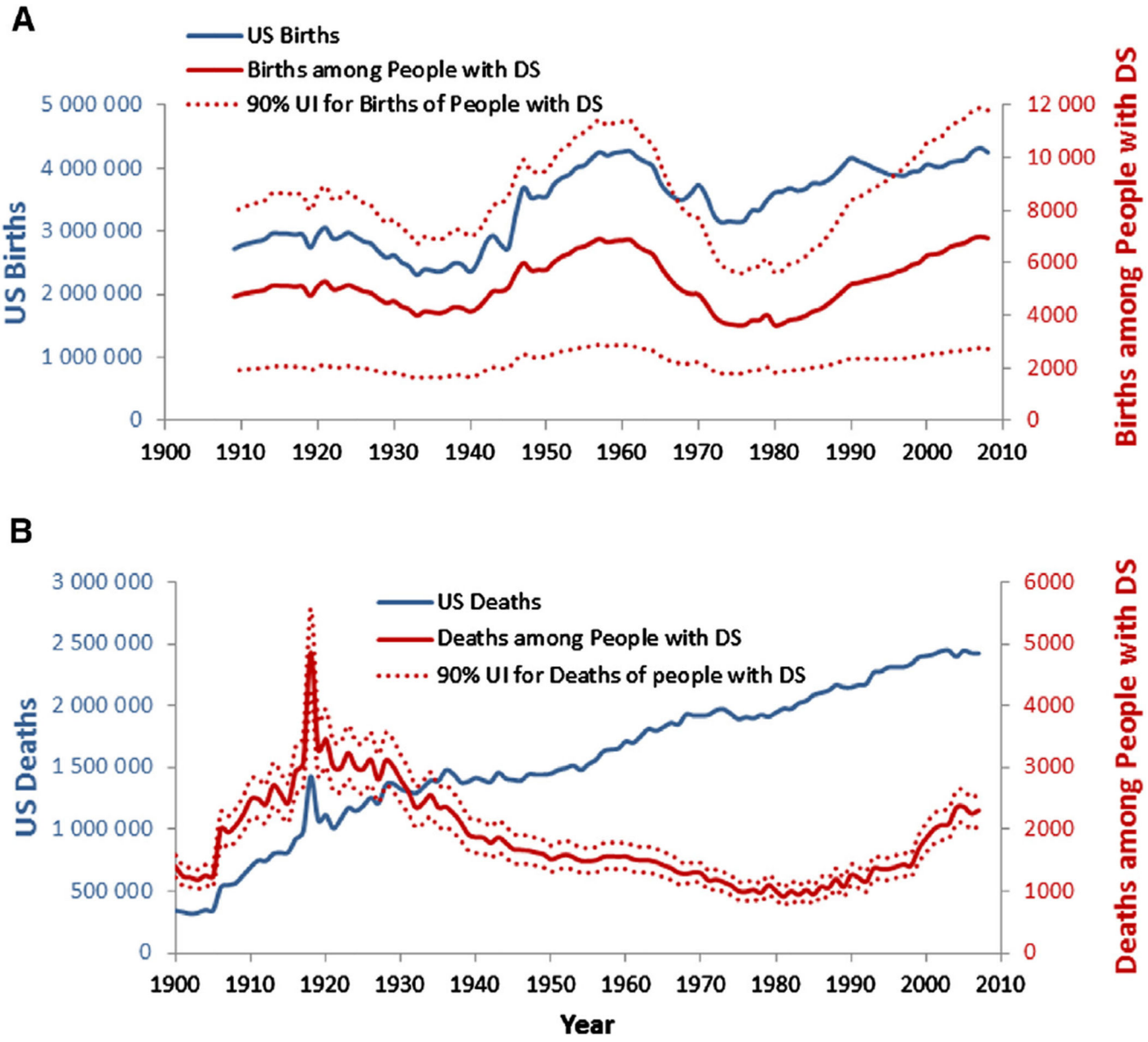

Figure 1.

Birth and death estimates for the US population and persons with Down syndrome for 1909–2007. In both panels, *blue* corresponds to the US population and *red* corresponds to the population of persons with Down syndrome. *Solid lines* indicate 50th percentile estimates from Monte Carlo sampling for both the US and Down syndrome data. *Dashed lines* indicate 90% UIs; UIs are absent from the US data because they are too narrow. **A**, Total births by year for 1909–2008. Although the US and Down syndrome curves are mostly parallel, there is a notable dip in the births of infants with Down syndrome in the 1970s owing to fewer births among women in their 30s and 40s. Both curves show an increase in births during the 1946–1964 baby boom, followed by a decline in the 1970s. This decline was primarily among older women, possibly related to the advent of the birth control pill in 1960, which enabled them to prevent additional pregnancies. Fewer births among older

women translated to even fewer births of infants with Down syndrome. **B**, Total deaths by year for 1900–2007. There is a spike in both populations in 1918, corresponding to the international influenza pandemic. *DS*, Down syndrome.

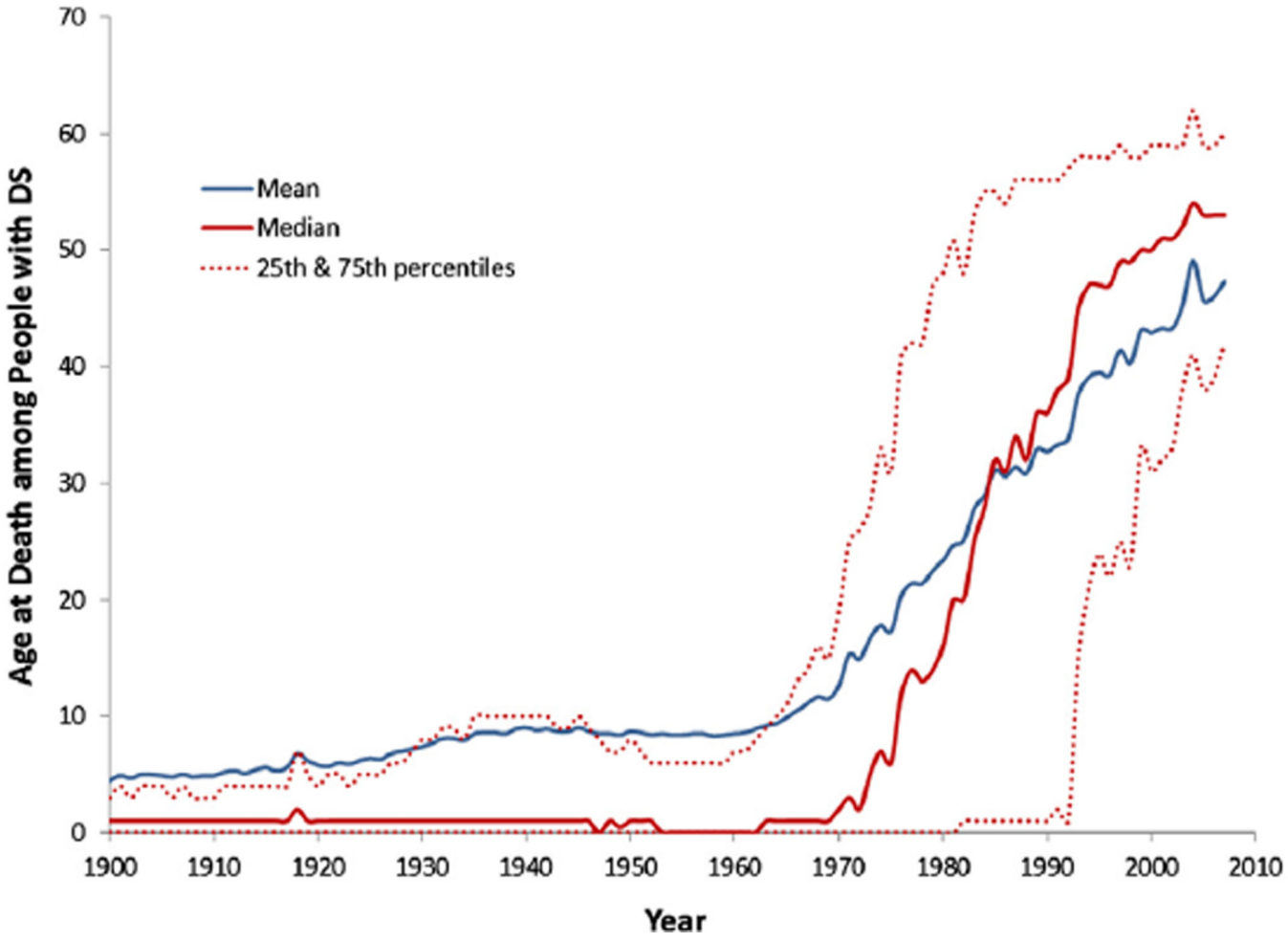

Figure 2.

Mean, median, and 25th and 75th percentiles for age at death in persons with Down syndrome, 1900–2007. The mean and median age at death for persons with Down syndrome have increased significantly over the past 40 years. In 2007, the mean and median ages at death were 47.3 and 53 years, respectively, reflecting a 3.75–fold increase in average life expectancy since 1970.

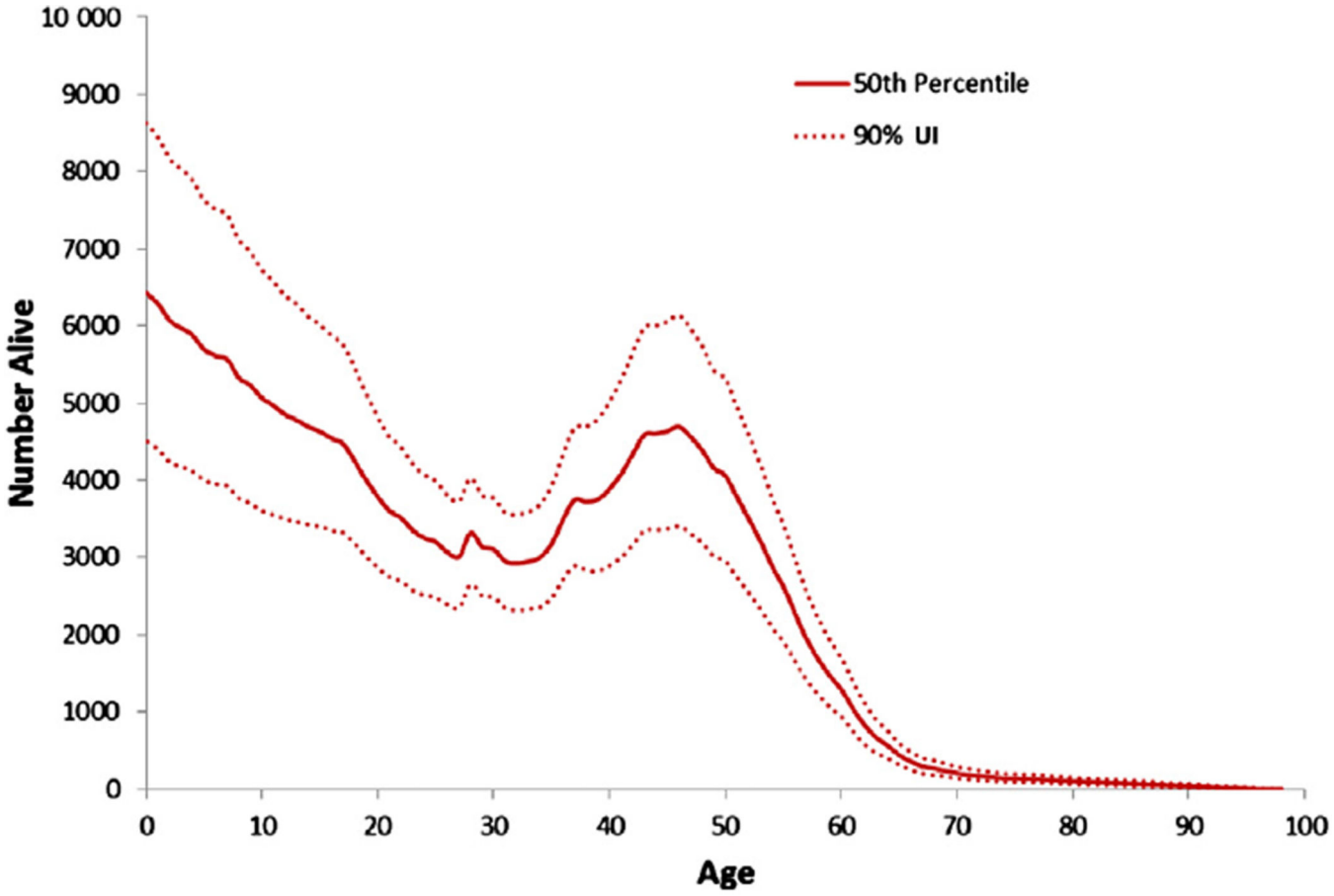

Figure 3.

Number of persons with Down syndrome in the US on January 1, 2008, by age. Estimates for the numbers of persons with Down syndrome alive at January 1, 2008, are plotted by their age. The *solid curve* indicates the 50th percentile estimate, and the *dashed curves* are 90% UIs (5th and 95th percentiles). There is a notable increase in the number of persons aged 35–60 years (born in 1947–1972), which can be explained by the 1946–1964 baby boom and the increased life expectancy in older individuals with Down syndrome.